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LETTER TO THE EDITOR

Abdominal pain in a patient with acute lymphoblastic leukaemia

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Dear Editor,

A 57-year-old man was admitted to our hospital with acute lower abdominal pain and diarrhoea. The patient had been diagnosed with a CD20 and *bcr-abl* positive B-cell acute lymphoblastic leukaemia 15 months ago and was treated with four cycles rituximab-Hyper-CVAD. He was healthy with a complete molecular remission at his last routine examination 4 weeks ago.

A CT scan of the abdomen revealed colitis of the colon descendens with perifocal oedema. Antibacterial therapy with piperazillin/tazobactam was initiated resulting in a normalisation of the initially elevated inflammatory blood markers. In contrast, the abdominal pain worsened although the analgesic therapy was substantially intensified. Due to increased pain, the patient's oral food intake was poor. Additionally, he developed tachycardia, hypertension as well as episodes with aggressiveness and loss of orientation. Further investigations, including extensive blood and bacterial stool analysis as well as a gastro- and colonoscopy, were unremarkable except for the urine, which turned from a clear yellow to a dark red colour (Fig. 1), highly suggestive for an acute porphyria. Indeed, increased levels of δ-aminolevulinic acid and porphobilinogen were found in the urine. All porphyrinogenic drugs were halted, an adequate caloric intake was guaranteed by nasogastric feeding and the patient was treated with exogenous heme-arginate for 4 days. The patient recovered completely from his symptoms within a few days and was discharged after 2 weeks of hospitalisation. A molecular analysis was positive for the presence of the W283X-mutation in the porphobilinogen deaminase gene, a common mutation in Swiss residents with acute intermittent porphyria (AIP). He was completely asymptomatic when last seen in April 2009.

AIP is an autosomal-dominant inherited disease of the heme biosynthetic pathway with a prevalence of symptomatic disease of one to two per 100,000 [1]. In AIP, the activity of the porphobilinogen deaminase (PBG) is reduced, resulting in an accumulation of upstream porphyrin precursors δ-aminolevulinic acid and porphobilinogen. At the moment, over 250 mutations in the PBG gene are known. Nearly 60% of all Swiss patients with an AIP carry the nonsense mutation W283X [2]. The majority of the mutation carriers are clinically healthy and exhibit symptoms only in situations when precipitating factors are present [3]. These include various drugs, poor energy intake during fasting as well as stress from illness and fever. Under these circumstances, the heme biosynthetic pathway gets activated and results in an excess accumulation of the porphyrin precursors δ-aminolevulinic acid and porphobilinogen due to the PGB enzyme deficiency. The key issues for acute therapy are to avoid drugs known to be porphyrinogenic, to promptly treat infections and to maintain an adequate caloric intake. Furthermore, intravenous heme derivates supporting the increased heme demand are the most effective treatment modalities for acute porphyria attacks [4].

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Fig. 1 The patient's red urine during the acute attack

It is interesting to realise that this patient was treated by an intensive chemotherapy regimen without the development of any symptoms. Currently, he is under maintenance treatment with dasatinib, a novel *bcr-abl* tyrosine kinase inhibitor, from which it is not yet known if it is tolerated by patients with acute porphyria.

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